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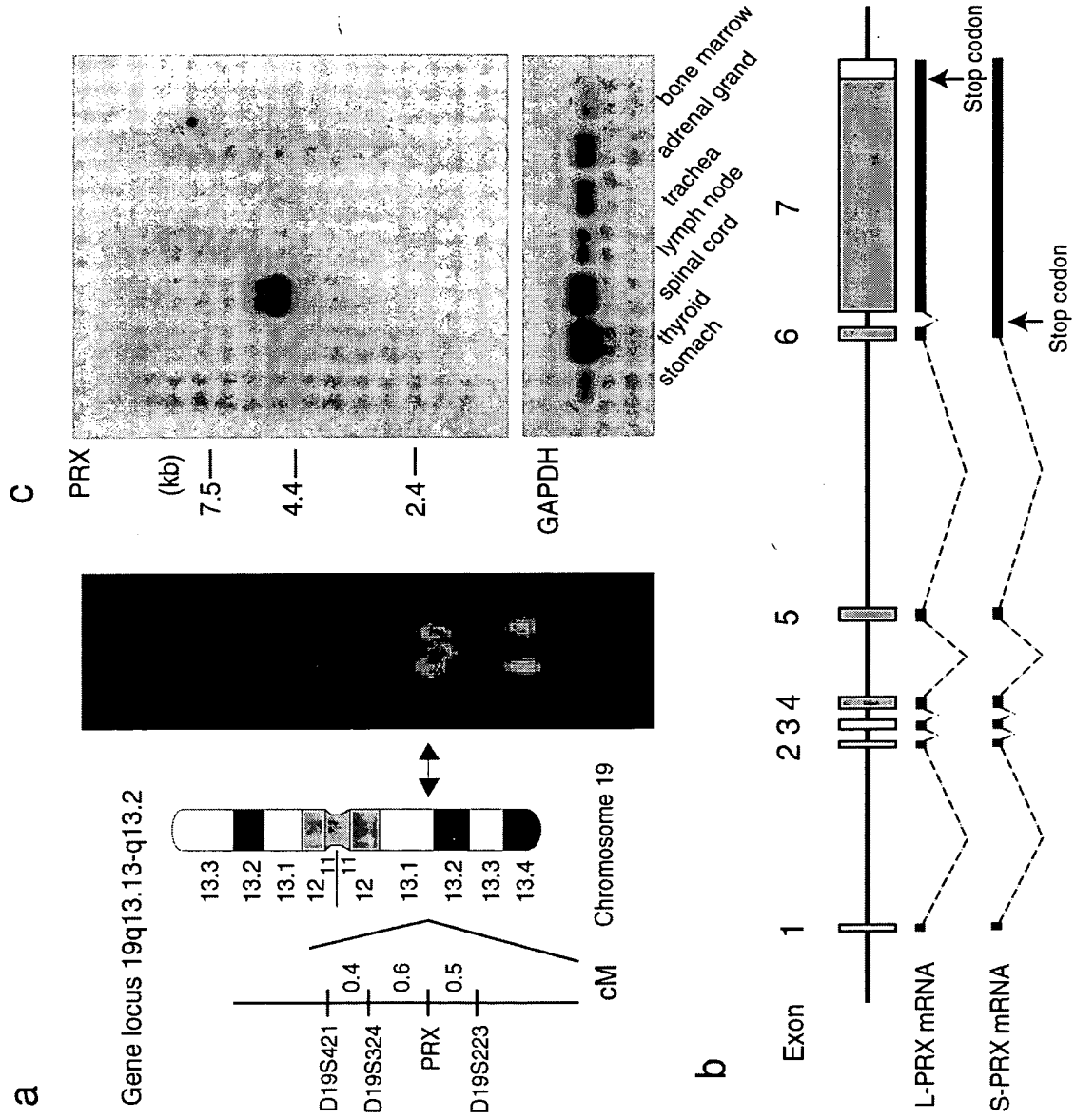


FIG. 1

PDZ Domain

Human	1	MEARS	SAEEL	RRAL	VEI	IV	ETE	TAQT	GVSG	IN	VAGG	GKEG	IF	VR	EL	REDS	PAAR	SL	SL	QEGD	QL	SAR	VFF	EN	FK	YED	AL	RL	L	QCA	EP	PK	VS	FL	CK	97																																																																																																																																																																																																																																																																																																																																																																																																															
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Rat	1	97																																																																																																																																																																																																																																																																																																																																																																																																															
Human		RTVPT	GD	LAL	RP	GT	VS	GYE	I	K	G	P	R	A	K	V	AK	LN	TQ	SL	S	P	V	K	K	K	K	K	K	K	K	K	K	K	K	197																																																																																																																																																																																																																																																																																																																																																																																																															
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Human		VAEEA	QAAR	LAAA	AP	PR	KAK	VEA	EA	GA	AR	FT	AP	QV	EL	VG	PR	LP	GA	EV	GP	VQ	VS	AP	KA	AP	SA	EA	AG	FAL	HL	PT	LGL	GAP	AP	PA	VG	IQ	PV	QV	297																																																																																																																																																																																																																																																																																																																																																																																																										
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Rat		297																																																																																																																																																																																																																																																																																																																																																																																																											
Human		PALPS	LPT	LPT	LP	CL	ET	REG	AV	SV	VVV	PT	L	D	VA	APT	V	GD	LAL	PG	AE	VE	AR	GE	AE	VAL	K	MP	RL	SF	PR	FG	AR	AE	VA	EAK	VAK	VS	PEAR	VK	GP	RL	MP	TF	397																																																																																																																																																																																																																																																																																																																																																																																																						
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Rat		397																																																																																																																																																																																																																																																																																																																																																																																																							
Human		LSLLE	PR	PA	AE	-	V	VE	SK	LKL	PT	IK	MP	SL	G	IG	VS	G	PE	VK	VP	K	GP	EV	KL	PK	AE	VE	KL	PK	VP	EA	AL	PE	VL	PE	VL	PE	VL	PE	VL	PE	VL	PE	VL	PE	VL	496																																																																																																																																																																																																																																																																																																																																																																																																			
Murine		497																																																																																																																																																																																																																																																																																																																																																																																																					
Rat		453																																																																																																																																																																																																																																																																																																																																																																																																					
Human		PKVSE	MKL	PKV	PE	MA	VP	EV	RL	PE	VQL	KV	SE	MKL	PKV	PE	MA	VP	EV	RL	PE	VQL	KV	SE	MKL	PE	VS	EA	VP	EV	RL	PE	VQL	PKV	PE	MA	VP	EV	RL	PE	VQL	PKV	PE	MA	VP	EV	RL	PE	VL	496																																																																																																																																																																																																																																																																																																																																																																																																	
Murine	

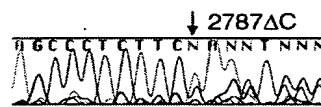
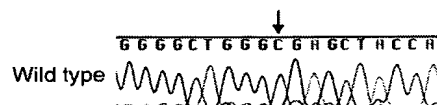
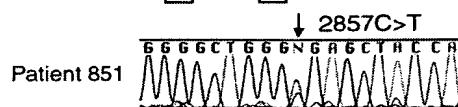
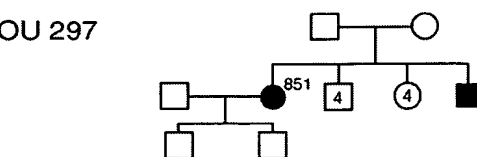
PDZ Domain

		PZD domain						
Human	1	M E A R S R S A E E L R R A E V I E T E A Q T G V S G I N V A G G K G E I F V R E L R E D P S A A R S L S L Q E G D Q L L S A R V F F E N F K Y E D A L R L L Q C A E P Y K V S F C L K	:	:	:	:	:	97
Murine	1	: : : : : : : : : : : : F : : : : : : : : K :	:	:	:	:	:	97
Rat	1	: : : : : : : : : : : : F : : : : : : : : K :	:	:	:	:	:	97
Human		R T V P T G D L A L R P G T V S G Y E I K G P R A K V A K L	:	:	:	:	:	147
Murine		: : : : : M : : : : : : : V : Q : S : : R : A : A :	:	:	:	:	:	148
Rat		: : : : : M : : : : ~ : : A : A : A : ~ :	:	:	:	:	:	147

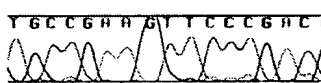
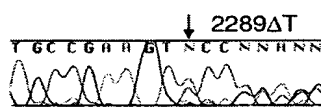
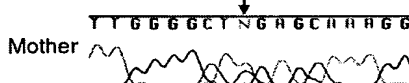
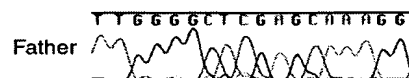
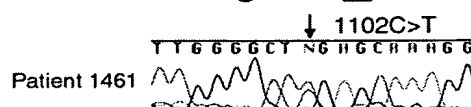
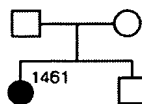
FIG. 2

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HOU 297



HOU 579



HOU 418

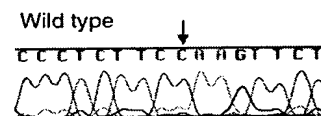
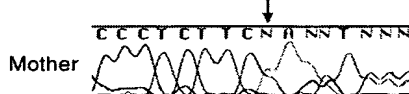
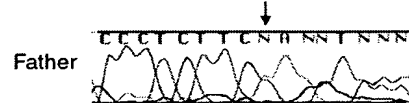
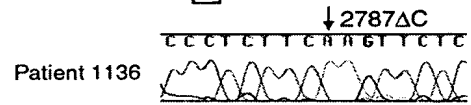
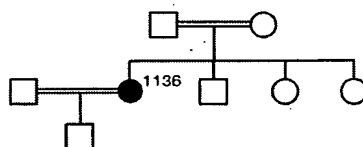
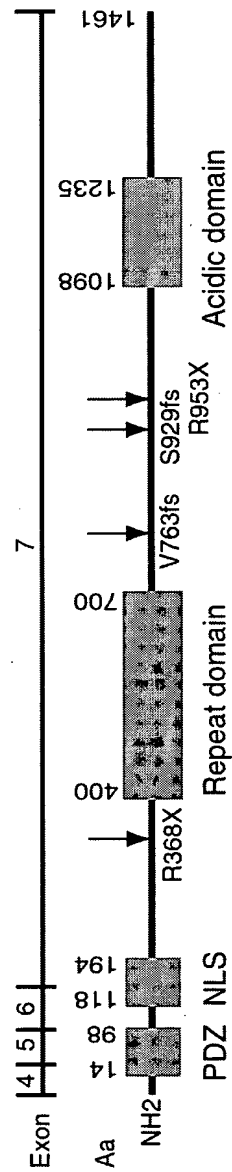


FIG. 3

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Patient	Phenotype	Pathogenic Allele 1	Frequency Allele 1	Pathogenic Allele 2	Frequency Allele 2
851	DSN	2857C>T R953X	0/190	2787ΔC S929fsX957	0/190
1461	DSN	1102C>T R368X	0/178	2289ΔT V763fsX774	0/176
1136	DSN	2787ΔC S929fsX957	0/190	2787ΔC S929fsX957	0/190

FIG. 4

Figure 5 shows two pedigree charts and corresponding DNA sequencing chromatograms. The top pedigree chart shows a family with four children (PN-44.1, PN-44.2, PN-44.3, PN-44.4) and their parents. The bottom pedigree chart shows a family with six children (PN-761.1, PN-761.2, PN-761.3, PN-761.4, PN-761.5, PN-761.6) and their parents. The DNA sequencing chromatograms show the results of sequencing for each individual, with specific mutations identified.

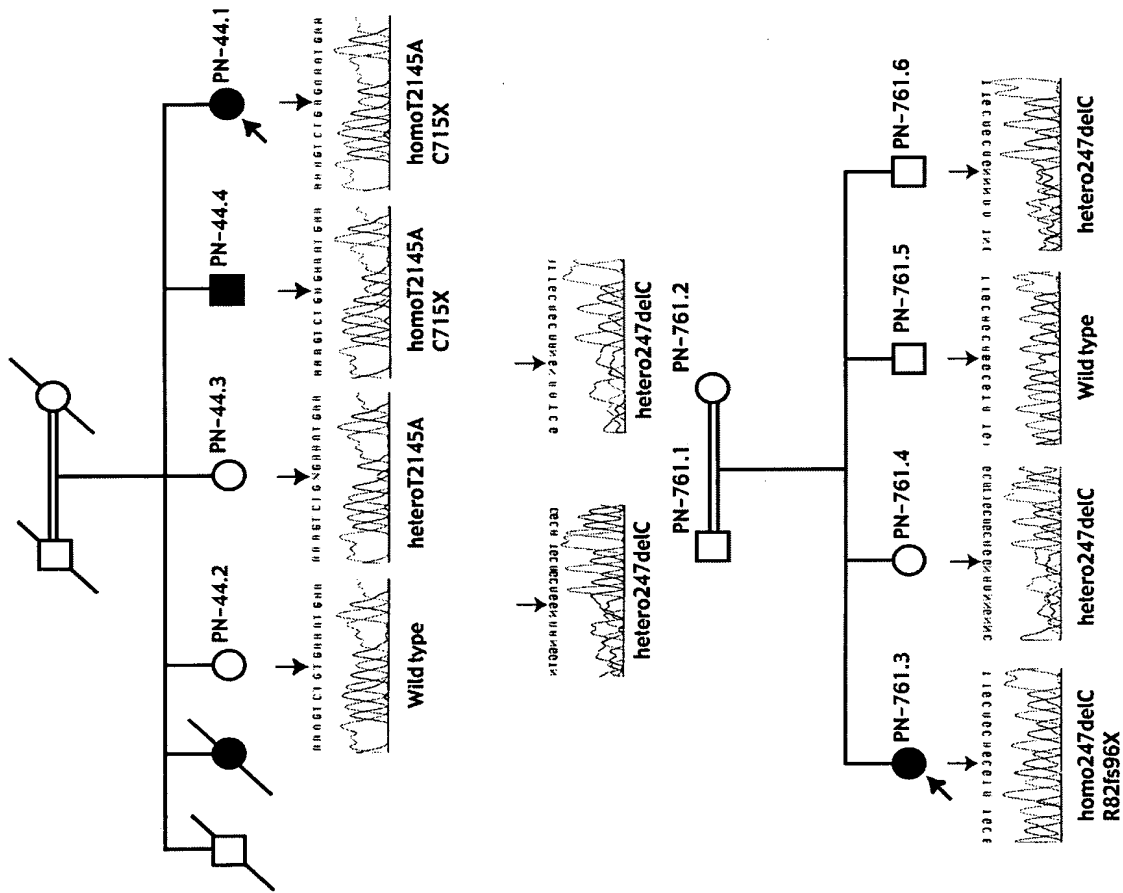


FIG. 5

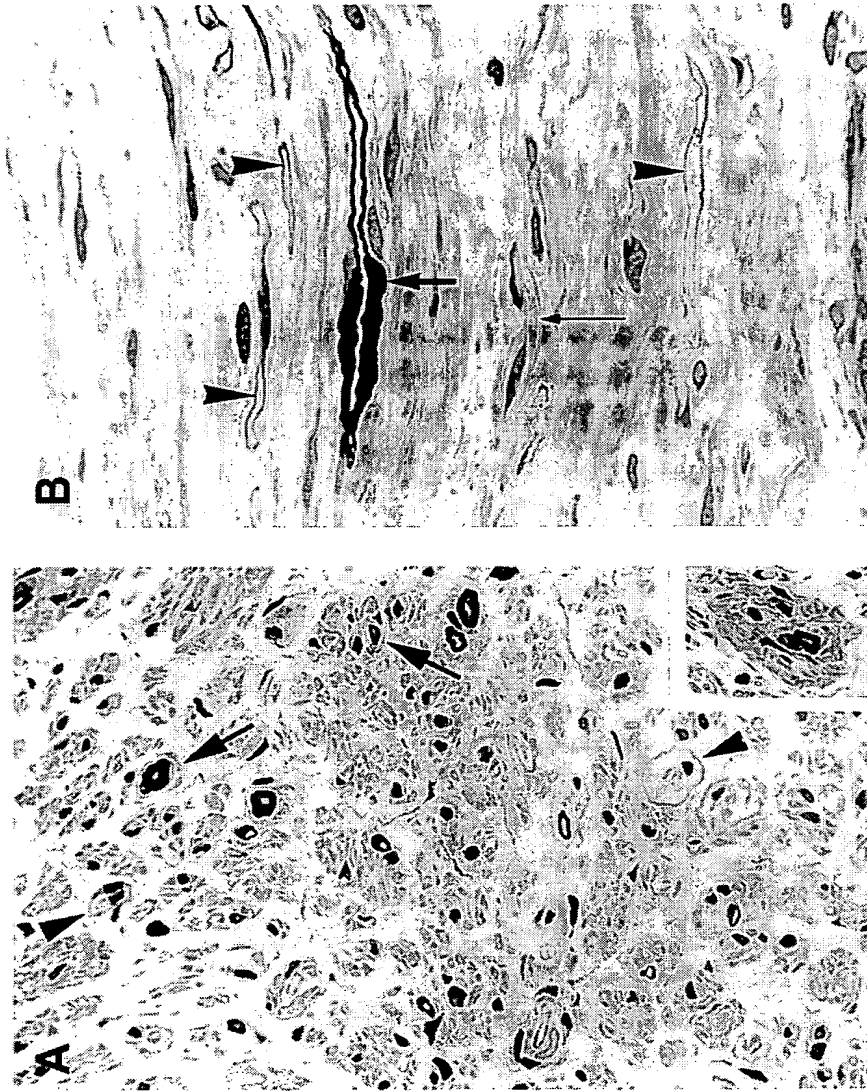


FIG. 6

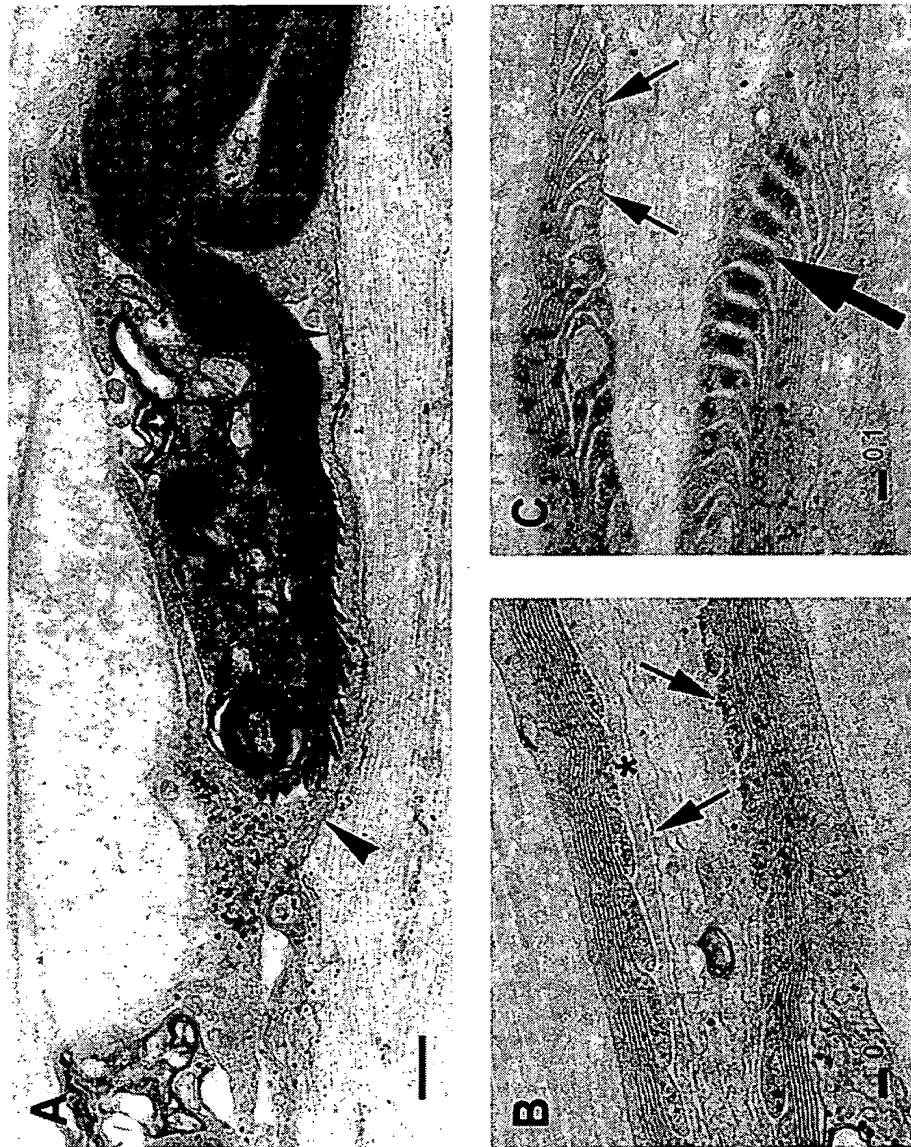


FIG. 7

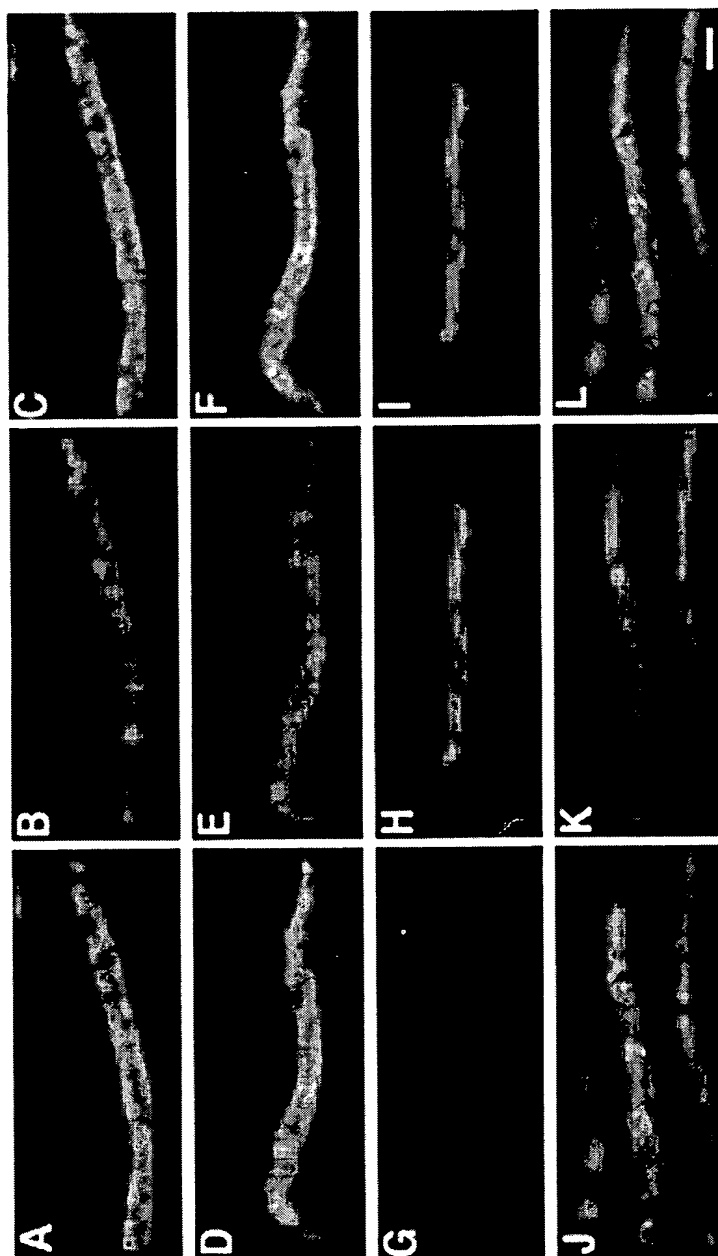
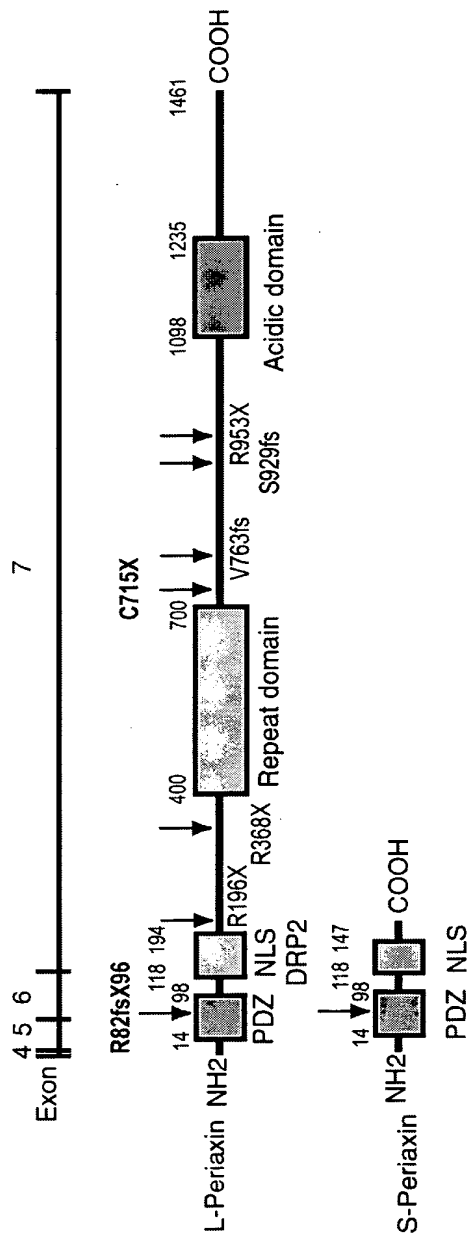


FIG. 8



Patient	Phenotype	Pathogenic Allele 1	Frequency	Pathogenic Allele 2	Frequency
PN44.1	CMT	C715X	0/180	C715X	0/180
PN44.4	CMT	C715X	0/180	C715X	0/180
PN761.3	CHN	R82fsX96	0/190	R82fsX96	0/190
851	DSN	R953X	0/190	S929fsX957	0/190
1461	DSN	R368X	0/178	V763fsX774	0/176
1136	DSN	S929fsX957	0/190	S929fsX957	0/190
CMT4F	DSN	R196X		R196X	

FIG. 9